# Developmental assessment in children and approach to developmental delay


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# What is normal development

- humans develop through a dynamique sequential process since the early days of life as embryo and the process continues after birth

# Child's development is a dynamic process

# Developmental domains include

**Gross motor**: Movements using the large muscles

**Fine motor/adaptive** : Movements using the hands and smaller muscles, often involving daily living skills

**Social/behavioural** : Attachment, self-regulation, and interaction with others

**Language** :Receptive and expressive communication, speech, and nonverbal communication

# American Academy of Pediatrics Policy on Developmental Screening

- All infants and young children should be screened for developmental delays
- Screening should be incorporated into the ongoing health care of the child
- Screening needs to be periodic
- Systematically eliciting parental concerns about development is an important new method
- Pediatricians should consider using standardized screening tool

The American Academy of Pediatrics (AAP) recommends that clinicians:

**screen** children for general development using **standardized,** validated tools at 9, 18, and 24 or 30 months

for autism at 18 and 24 months

**or at any point when** a caregiver or the clinician has a concern

# Screening Tests?

Detection rates without screening tests in pediatric practices

- **70%** of children with developmental disabilities not identified

- **80%** of children with mental health problems not identified

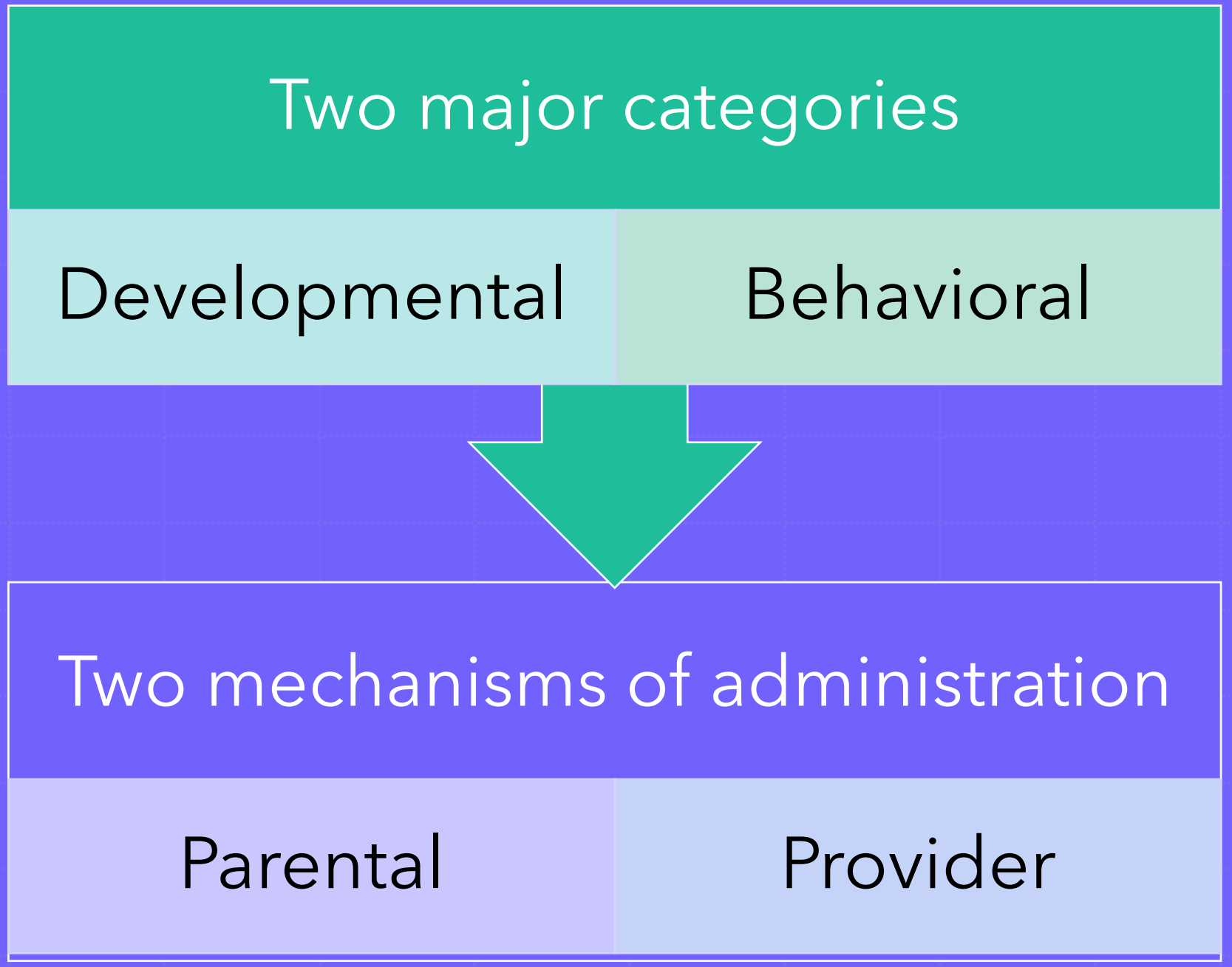
# Screening

Detection rates with screening tests in paediatric practices

- 70% to 80% of children with developmental disabilities correctly identified

- 80% to 90% of children with mental health problems correctly identified

# Screening tools



# Developmental screening tools

- Provider
- Denver
- CAT/CLAMS
- Bayley
- Brigance
- DIAL-R
- Parent
- Ages and Stages Questionnaire
- Parent's Evaluations of Developmental Status


- Emerging patterns of development

| Table 10-2 | Emerging Patterns of Behavior During the 1st Yr of Life* |
|---|---|
| **NEONATAL PERIOD (1ST 4 WK)** | |
| Prone: | Lies in flexed attitude; turns head from side to side; head sags on ventral suspension |
| Supine: | Generally flexed and a little stiff |
| Visual: | May fixate face on light in line of vision; "doll's-eye" movement of eyes on turning of the body |
| Reflex: | Moro response active; stepping and placing reflexes; grasp reflex active |
| Social: | Visual preference for human face |
| **AT 1 MO** | |
| Prone: | Legs more extended; holds chin up; turns head; head lifted momentarily to plane of body on ventral suspension |
| Supine: | Tonic neck posture predominates; supple and relaxed; head lags when pulled to sitting position |
| Visual: | Watches person; follows moving object |
| Social: | Body movements in cadence with voice of other in social contact; beginning to smile |
| **AT 2 MO** | |
| Prone: | Raises head slightly farther; head sustained in plane of body on ventral suspension |
| Supine: | Tonic neck posture predominates; head lags when pulled to sitting position |
| Visual: | Follows moving object 180 degrees |
| Social: | Smiles on social contact; listens to voice and coos |
| **AT 3 MO** | |
| Prone: | Lifts head and chest with arms extended; head above plane of body on ventral suspension |
| Supine: | Tonic neck posture predominates; reaches toward and misses objects; waves at toy |
| Sitting: | Head lag partially compensated when pulled to sitting position; early head control with bobbing motion; back rounded |
| Reflex: | Typical Moro response has not persisted; makes defensive movements or selective withdrawal reactions |
| Social: | Sustained social contact; listens to music; says "aah, ngah" |
| **AT 4 MO** | |
| Prone: | Lifts head and chest, with head in approximately vertical axis; legs extended |
| Supine: | Symmetric posture predominates, hands in midline; reaches and grasps objects and brings them to mouth |
| Sitting: | No head lag when pulled to sitting position; head steady, tipped forward; enjoys sitting with full truncal support |
| Standing: | When held erect, pushes with feet |
| Adaptive: | Sees raisin, but makes no move to reach for it |
| Social: | Laughs out loud; may show displeasure if social contact is broken; excited at sight of food |

| | |
|---|---|
| **AT 7 MO** | |
| Prone: | Rolls over; pivots; crawls or creep-crawls (Knobloch) |
| Supine: | Lifts head; rolls over; squirms |
| Sitting: | Sits briefly, with support of pelvis; leans forward on hands; back rounded |
| Standing: | May support most of weight; bounces actively |
| Adaptive: | Reaches out for and grasps large object; transfers objects from hand to hand; grasp uses radial palm; rakes at raisin |
| Language: | Forms polysyllabic vowel sounds |
| Social: | Prefers mother; babbles; enjoys mirror; responds to changes in emotional content of social contact |
| **AT 10 MO** | |
| Sitting: | Sits up alone and indefinitely without support, with back straight |
| Standing: | Pulls to standing position; "cruises" or walks holding on to furniture |
| Motor: | Creeps or crawls |
| Adaptive: | Grasps objects with thumb and forefinger; pokes at things with forefinger; picks up pellet with assisted pincer movement; uncovers hidden toy; attempts to retrieve dropped object; releases object grasped by other person |
| Language: | Repetitive consonant sounds ("mama," "dada") |
| Social: | Responds to sound of name; plays peek-a-boo or pat-a-cake; waves bye-bye |
| **AT 1 YR** | |
| Motor: | Walks with one hand held; rises independently, takes several steps (Knobloch) |
| Adaptive: | Picks up raisin with unassisted pincer movement of forefinger and thumb; releases object to other person on request or gesture |
| Language: | Says a few words besides "mama," "dada" |
| Social: | Plays simple ball game; makes postural adjustment to dressing |

| | |
|---|---|
| **15 MO** | |
| Motor: | Walks alone; crawls up stairs |
| Adaptive: | Makes tower of 3 cubes; makes a line with crayon; inserts raisin in bottle |
| Language: | Jargon; follows simple commands; may name a familiar object (e.g., ball); responds to his/her name |
| Social: | Indicates some desires or needs by pointing; hugs parents |
| **18 MO** | |
| Motor: | Runs stiffly; sits on small chair; walks up stairs with 1 hand held; explores drawers and wastebaskets |
| Adaptive: | Makes tower of 4 cubes; imitates scribbling; imitates vertical stroke; dumps raisin from bottle |
| Language: | 10 words (average); names pictures; identifies 1 or more parts of body |
| Social: | Feeds self; seeks help when in trouble; may complain when wet or soiled; kisses parent with pucker |
| **24 MO** | |
| Motor: | Runs well, walks up and down stairs, 1 step at a time; opens doors; climbs on furniture; jumps |
| Adaptive: | Makes tower of 7 cubes (6 at 21 mo); scribbles in circular pattern; imitates horizontal stroke; folds paper once imitatively |
| Language: | Puts 3 words together (subject, verb, object) |
| Social: | Handles spoon well; often tells about immediate experiences; helps to undress; listens to stories when shown pictures |

**30 MO**

| | |
|---|---|
| Motor: | Goes up stairs alternating feet |
| Adaptive: | Makes tower of 9 cubes; makes vertical and horizontal strokes, but generally will not join them to make cross; imitates circular stroke, forming closed figure |
| Language: | Refers to self by pronoun "I"; knows full name |
| Social: | Helps put things away; pretends in play |

**36 MO**

| | |
|---|---|
| Motor: | Rides tricycle; stands momentarily on 1 foot |
| Adaptive: | Makes tower of 10 cubes; imitates construction of "bridge" of 3 cubes; copies circle; imitates cross |
| Language: | Knows age and sex; counts 3 objects correctly; repeats 3 numbers or a sentence of 6 syllables; most of speech intelligible to strangers |
| Social: | Plays simple games (in "parallel" with other children); helps in dressing (unbuttons clothing and puts on shoes); washes hands |

| | |
|---|---|
| **48 MO** | |
| Motor: | Hops on 1 foot; throws ball overhand; uses scissors to cut out pictures; climbs well |
| Adaptive: | Copies bridge from model; imitates construction of "gate" of 5 cubes; copies cross and square; draws man with 2-4 parts besides head; identifies longer of 2 lines |
| Language: | Counts 4 pennies accurately; tells story |
| Social: | Plays with several children, with beginning of social interaction and role-playing; goes to toilet alone |
| **60 MO** | |
| Motor: | Skips |
| Adaptive: | Draws triangle from copy; names heavier of 2 weights |
| Language: | Names 4 colors; repeats sentence of 10 syllables; counts 10 pennies correctly |
| Social: | Dresses and undresses; asks questions about meaning of words; engages in domestic role-playing |

# Vision

- Visual fixation : by 35weeks of gestation fixation is present
- 2 months :follows objects to 180 degree
- Preference to human faces
- Visual acuity : very low , reaches approximate adult levels by 1 year of age
- Accommodation + convergence :2-3 months

# Vision

- Visual field : approximate adult level by 1 year of age
- saccade : since birth , but slower than in adults
- Categorization of colors : since birth
- Categorization of orientation : since birth
- The visual system reaches its full maturity at the age of 10-11 years

# Hearing

Development of hearing : excellent since birth (since 5th month of fetal life )

In the first 10 months of life : hearing is the dominant sense , after this period visual sense predominates

# Hearing

Hearing assessment : difficult in infants

The examiner must be behind the infant

The infant turns to the source of noise by the age of 3 months

**but this test is neither accurate nor constant : ie** if the infant doesn't turn to the voice it is not necessarily that he doesn't hear. The test is variable from one moment to another

# Gross motor function –first year of life

- Neonate : moves head from side to side
- 1 m:raises head momentarily when prone
- 2m:raises head (sustained ) when prone
- 3m: raises head above plane of body

- 4m: supports weight , full head control, sits with full truncal support


- 7m: sits with pelvic support, rolls over
- 10m :sits alone, crawl , pulls to standing position ,walks holding in furniture
- 12m: walks with hand held
- 15m : walks alone, crawls upstairs
- 18m :runs stiffly , walks upstairs one hand held
- 24m=2years : runs well , up+ downstairs one step at a time

- 2.5years : upstairs alternating feet
- 3years : rides tricycle , stands momentarily on 1 foot
- 4 years : hops on 1 foot , throws ball
- 5 years : skips


# Fine motor

# Fine motor

- 3m : opens hand spontaneously , reach objects
- 4 m : reaches towards objects+ put in mouth
- 6m-7 : transfer from hand to hand
- 10m : thumb index grasp, releases objects grasped by others
- 12m : releases object on demand

# Fine motor

- 15m :3 cubes
- 1.5years :4 cubes , imitates vertical stroke
- 2 years 7 cubes , imitates horizontal stroke
- 2.5 years : 9 cubes , makes vertical + horizontal stroke
- 3years : circle
- 4 years : cross , square , draw a man test

# Language

3m : aah , ngah

7m : pollysylabisc vowels

10m :few words besides mama , dada

15m:follows simple commands , may name a familiar object

18m: 10 words , names pictures , identifies parts of body

2 years : put 3 words together

4years :tells a story , count 4 pennies correctly

5 years :counts 10 pennies correctly

# Social

- Neonate : visual preference to human face
- 1 m : beginning to smile
- 2m : social smile , listen to voice + coos
- 3 m : sustained social contact
- 4m : laughs loudly, excited at sight of food
- 7m :enjoys mirror , prefers mother
- 10m : waves bye , plays peek a boo , responds to sound of name

1 year : plays simple boy game , postural adjustment to dressing

15m : hugs parents , indicates desires by pointing

1.5 year :feeds self , kisses parents with pucker

2 years : listen to stories with pictures

2.5 years : pretend in play

3 years : plays simple games with children , washes hands unbutton clothing , put on shoes

4 years :goes to toilet alone , plays with several children ( role playing )

5 years : dresses + undresses , asks questions about meaning of words

# Red flags worrisome if still not reached

babbling by 12 months

gesturing (e.g., pointing, waving bye-bye) by 12 months

single words by 16 months

two-word spontaneous (not just echolalic) phrases by 24 months

loss of any language or social skills at any age

# Videos : how to perform developmental assessment

- https://mrcpch.paediatrics.co.uk/development/development-videos/

## Developmental delay

Child not reaching developmental milestones at the expected age , even after allowing the broad variation of normality

Global developmental delay : when ≥2 domains in development are delayed

The earlier the identification of these children the better outcome( birth -2years )

Favorable environment: enhances + optimize brain development

# Consequences of Early Childhood Developmental Problems

- Low self-esteem
- Poor relationship formation
- Poor academic success
- Conduct problems
- Truancy and school drop-out
- Unemployment
- Poor quality parenting skills

# Developmental surveillance

5-10% of pediatric population have developmental disabilities

To identify these children : developmental **surveillance**:

1-observe infant

2-take developmental history

3-ellicit parental concerns

Developmental **screen** : necessary adjunct

# Developmental delay

Developmental delay :Slow progress in the attainment of developmental milestones

Psychomotor regression: loss of developmental milestones previously attained.

# Two important questions to answer

Is developmental delay restricted to specific areas or is it global ( 2 or more )?

Is  it development delayed or is child regressing

Determine is the delay static or progressive

*Predominant speech delay*

- Hearing impairment
- Autism
- Bilateral hippocampal sclerosis
- Congenital bilateral perisylvian syndrome

# Motor delay

Ataxia

Hemiplegia

paraplegia

Hypotonia

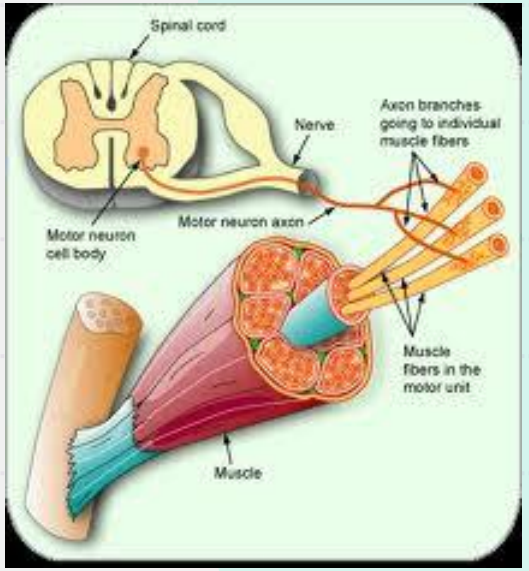
Neuromuscular disorders

Determine if the motor delay is due to central ( upper motor neurone disorder ) or peripheral ( lower motor neurone disorder

**Or combined** : example Duchenne muscle dystrophy ,Cong muscle dystrophies ,Metachroamtic leukodystrophy

# Peripheral

# Central




# Differentiating central from peripheral causes: hx and ex

- Normal or **Brisk reflexes**
- Other abnormal brain functions: delay, seizures
- Fisting

- Scissoring on vertical suspension
- Dysmorphic features
- Extra-cranial organ malformations

- **Absent** or Depressed reflexes
- Intact brain function
- Awake and alert
- Muscle atrophy
- Profound **weakness**
- Fasciculations
- No extra-cranial organ malformations

# *Global developmental delay ( 2 or more) :examples*

- Perinatal insult : asphyxia , congenital infections, bilirubin toxicity
- Chromosomal /genetic
- Metabolic : hypothyroidism , neurometabolic disorders
- Cerebral malformations
- Progressive neurodegenerative disorders

- Determine is it static or progressive
- Age of onset of symptoms
- Clues in history and examination

# Evaluation of the child with global developmental delay

# Look for the hints

- History :antenatal, birth history , past history
- Age of Onset of symptoms :static or progressive
- Family history ( very detailed ):consanguinity, similar cases , deaths in early infancy
- Examination : dysmorphysm
- systems involvement
- Results of investigations

# Clues in the history

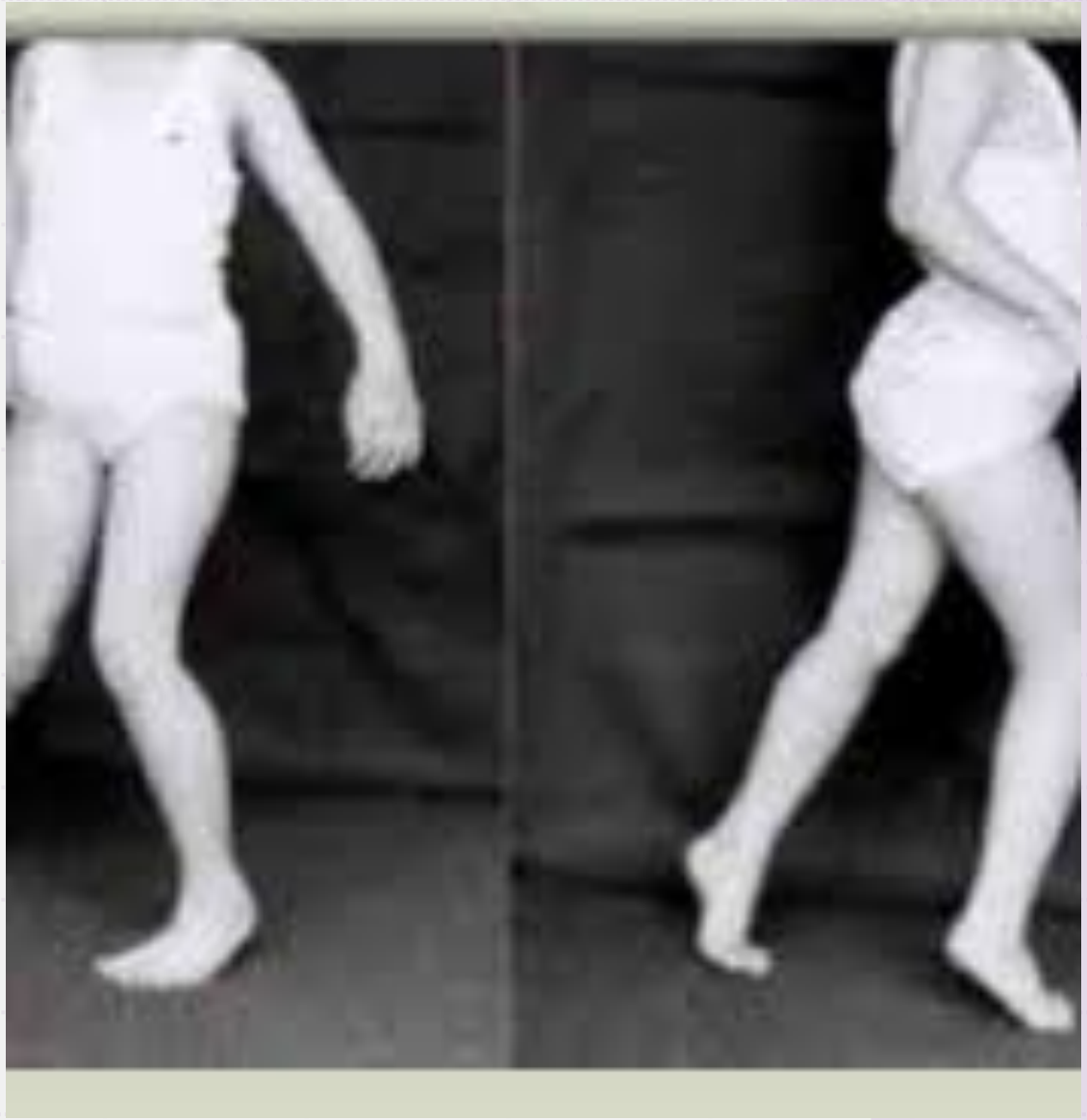
Antenatal history : trauma , infection, death of a twin , hypertension , diabetes …

Birth history : weight , gestational age hyperbilirubinemia ..

Past : meningitis , encephalitis , epilepsy trauma …

# What is your most likely dx based on the clues

- 20 months old boy presented with history of delayed walking , he also has weakness of the right side of body
- Antenatal history revealed decreased fetal movement , he was born prematurely at 28 weeks of gestation



# Cerebral palsy: definition

non progressive (static) disorder of motor function and movement that usually manifests early **in** life as a result of central nervous system damage to the developing brain

Incidence :1.5 to 2.5 per 1,000 live births

Most patients are identified by 2 years of age due to delayed motor milestones

More common **in children** who are born very prematurely or at term

Slightly higher prevalence **in** males M:F= 1.5:1

Poor prenatal care may increase the incidence of **cerebral palsy**

# Onset

- Prenatal
- Perinatal
- Post natal

# Causes

In most cases, the exact cause is unknown but is most likely multifactorial

**Majority of cases : not caused by hypoxic ischemic incidents occurring perinatally (as it was believed until recently.)**

**70-80% cases are prenatal in origin**

**Although prematurity is the most common known antecedent of CP, the majority of children who develop CP are born at term.**

# Causes

70% - 80% of cases of CP are due to **antenatal factors**

10% - 28% of cases are due to birth asphyxia **in** term and near-term infants

More than 1 etiologic factor is often identified.

The term CP is descriptive : different

etiologies and clinical presentations

1- According to the extremities involved

2-According to the characteristics of neurologic dysfunction.

3- Functional classification

# Hemiplegic CP

Arm > leg

walks : tip toes , swing the affected leg ( semicircular arc )

Corticosensory impairment: common

Mental retard. : 1/3

Seizures : 1/3

vision

# Spastic quadreplegic CP

- Generalized increase in muscle tone
- Legs > arms
- Opisthotonic posture ( first year of life )
- Difficulties in swallowing and articulation
- Incoordination of oropharyngal m :recurrent pneumonia
- Seizures :50%
- Mental retardation : majority
- Auditory , visual abn : common

# Diplegic

- Bilateral leg involvement
- Commonly *some* degree of UL involvement
- Infant : scissoring , older child : tip toewalk
- Seizures , MR , visual abn

# According to neurological dysfunction:

- Spastic: most common type ( 70%-80%)
- Ataxic: with cerebellar involvement
- Dyskinetic (extrapyramidal, choreoathetoid): due to predominant basal ganglia involvement **in** patients with acute severe hypoxia and kernicterus. Symptoms consistent with a movement disorder may appear later **in** life
- Mixed

# Diagnosis : HX + EX

Diagnosis : always a motor deficit

usual presentation :delayed  motor milestones

hand preference < 3 years: relative weakness of 1 side.

History: the child is not losing function= the patient does not have a progressive disease.

Examination  : hypotonia , spasticity, persistent primitive reflexes , underdevelopment of parachute reflex

# Associated conditions

- Mental retardation :30- 50%
- Ophthalmologic defects : 30%
- Hearing impairment : 10%
- Speech and language disorders :40%
- Epilepsy  : 30- 40 %

# Mangement and ttt

Multidisciplinary team

goal for the treatment program : maximize function + optimize development = help them participate in as many activities as possible in multiple social settings

# Types of interventions

- Physical therapy
- Orthopedic surgery (later)
- Muscle tone management
- orthosis

What is your diagnosis ? Clues in the family history

A 4 year old boy presented with history of global developmental delay , epilepsy and microcephaly

Parents are cousins , he has one cousin who was diagnosed to have phenylketonuria by neonatal screening

# phenylketonuria

Error of amino acids metabolism

Autosomal recessive

No acute clinical symptoms

Untreated leads to mental retardation

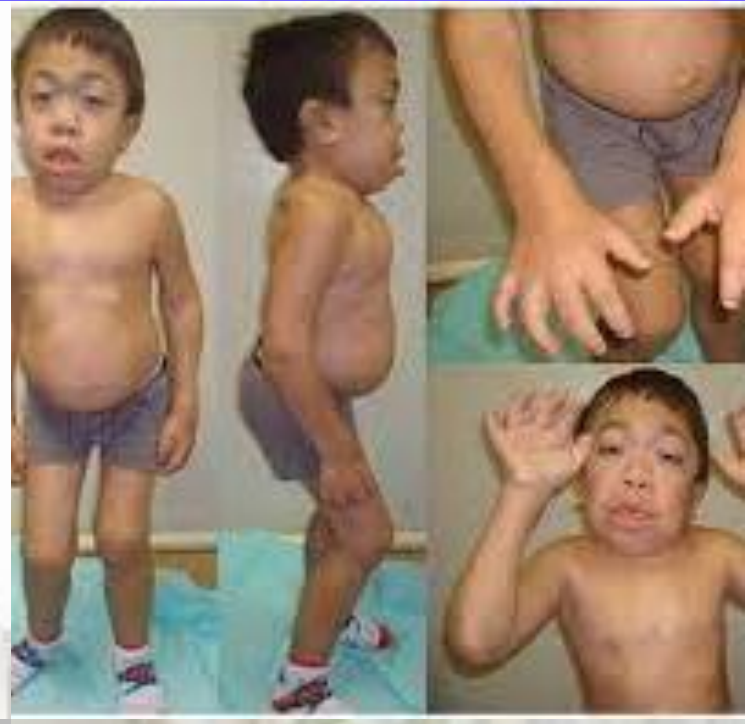
Associated complications: behavior disorders, cataracts, skin disorders, and movement disorders

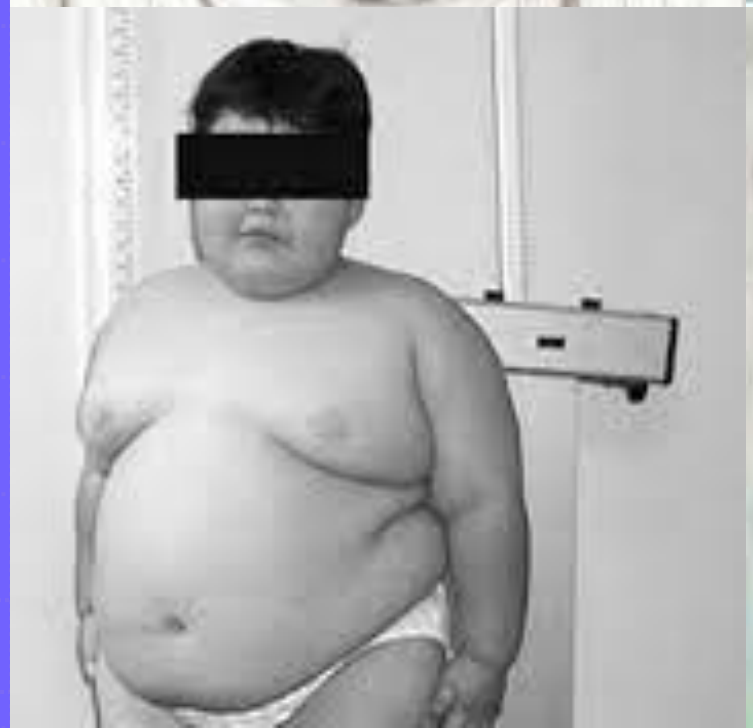
First newborn screening test was developed in 1959

Treatment: phenylalaine restricted diet (specialized formulas available)

# What is your diagnosis :

- Clues in the face

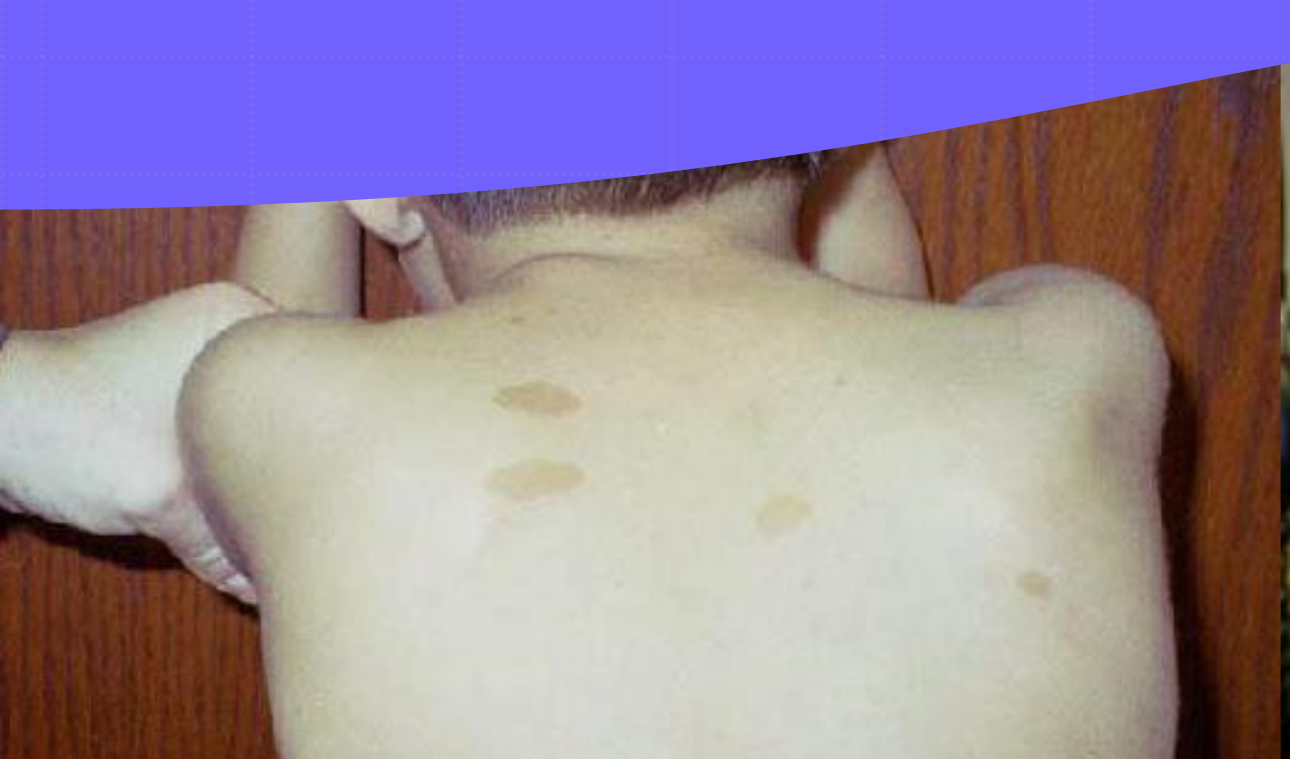


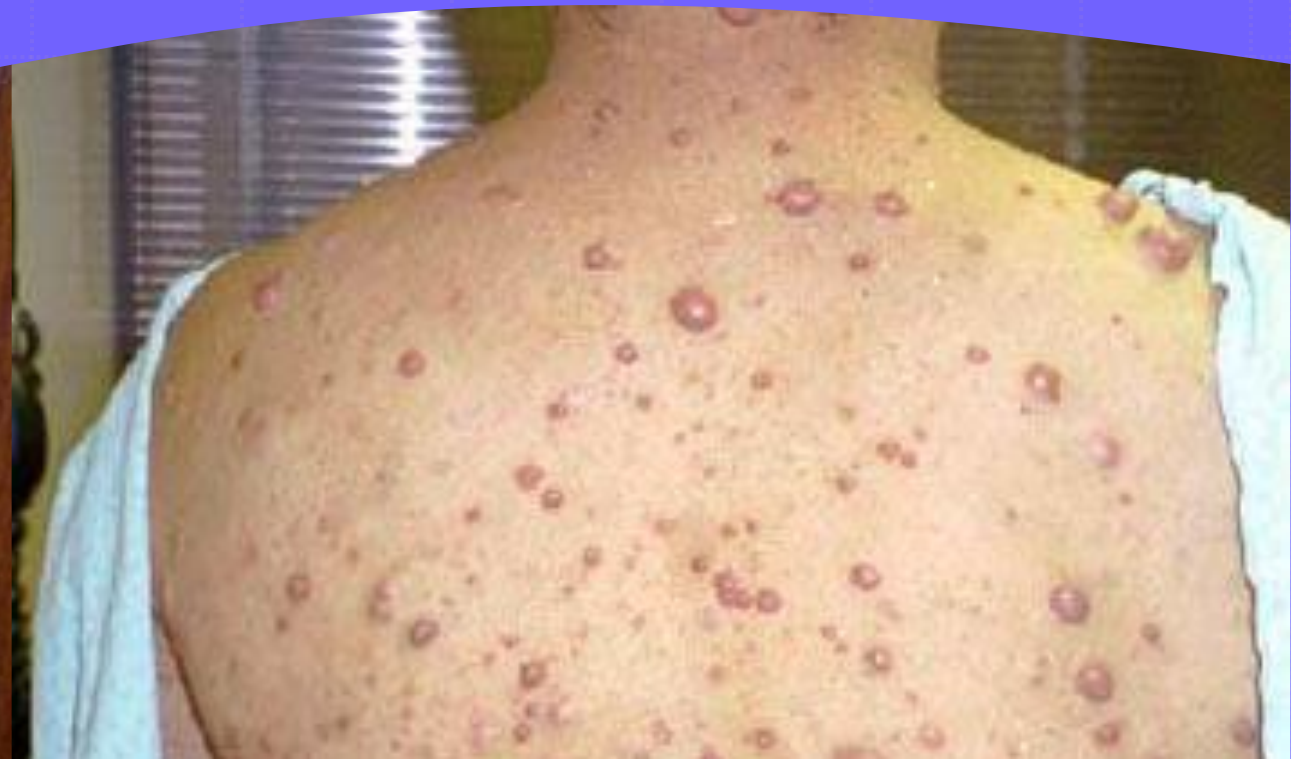


# Clues : skin

- Neurocutaneous disorders : examples
- Neurofibromatosis type 1
- Sturge weber syndrome
- Tuberous scleoris

# Clues in skin ??





# Neurofibromatosis type 1

Autosomal dominant with variable expression

Most common neurocutaneous syndrome

Diagnosis: dx criteria

Macrocepahly is common

Learning disability

Risk of neoplastic disorders

Seziures

Treatment : supportive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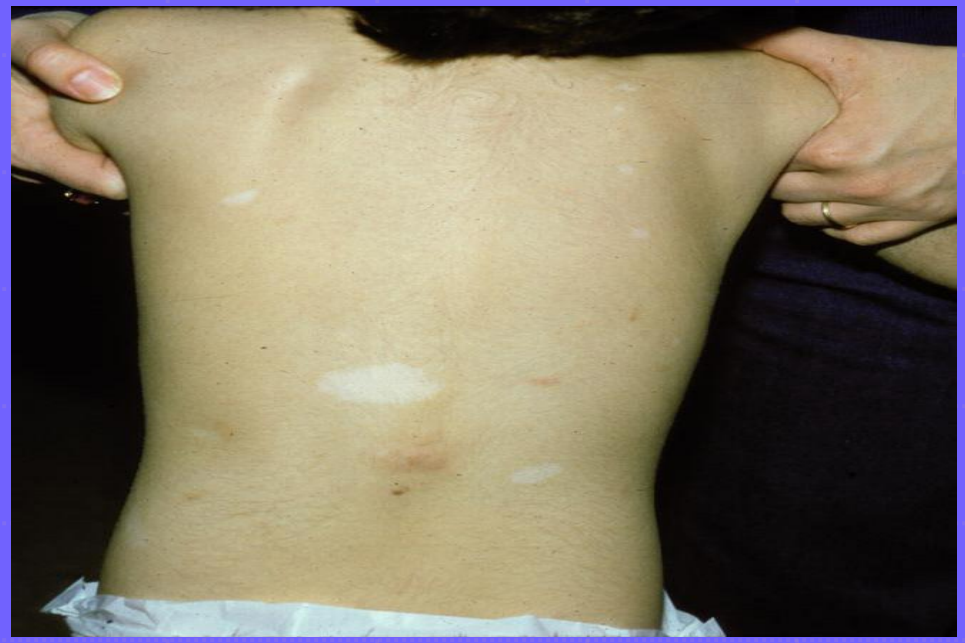
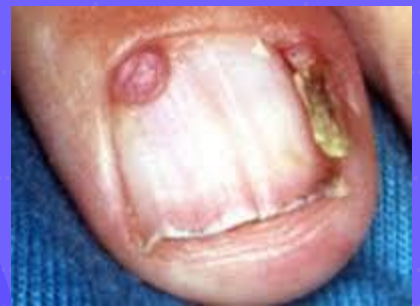




# Tuberous sclerosis

Autosomal dominant

2 genes ( TSC1 and TSC2 )

Characteristic skin lesions ( ash leaf , shagreen patches , achromic spots , sebaceous adenoma ….)

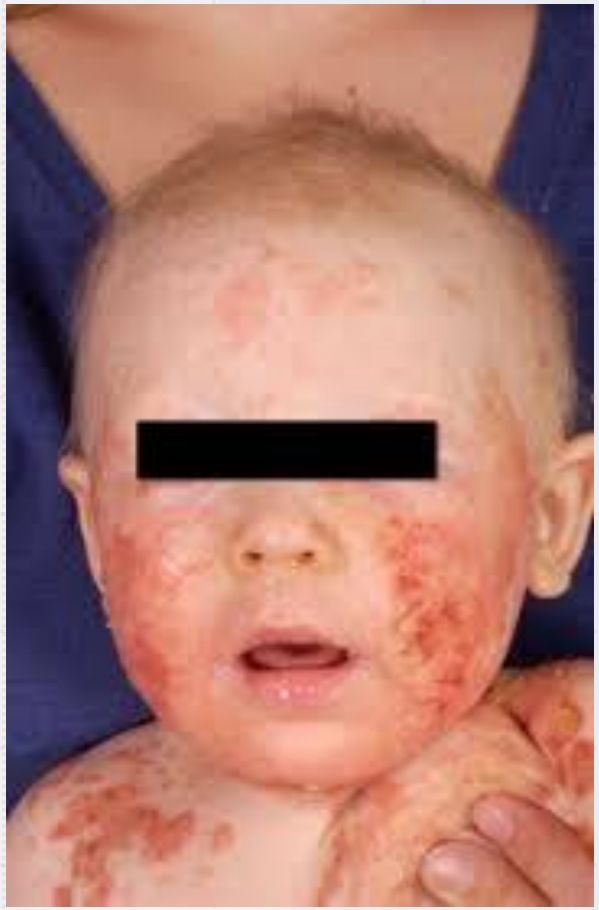
Epilepsy , developmental delay

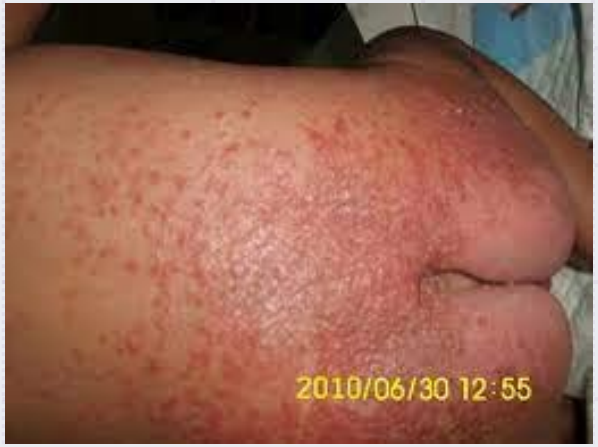
Subependymal hamartomas

Dx : dx criteria

# clues in skin : rash, dermatitis

- Biotinidase deficiency
- Propionic acidemia(orgnic acidemia)
- Refsum disease(peroxysomal disorder)

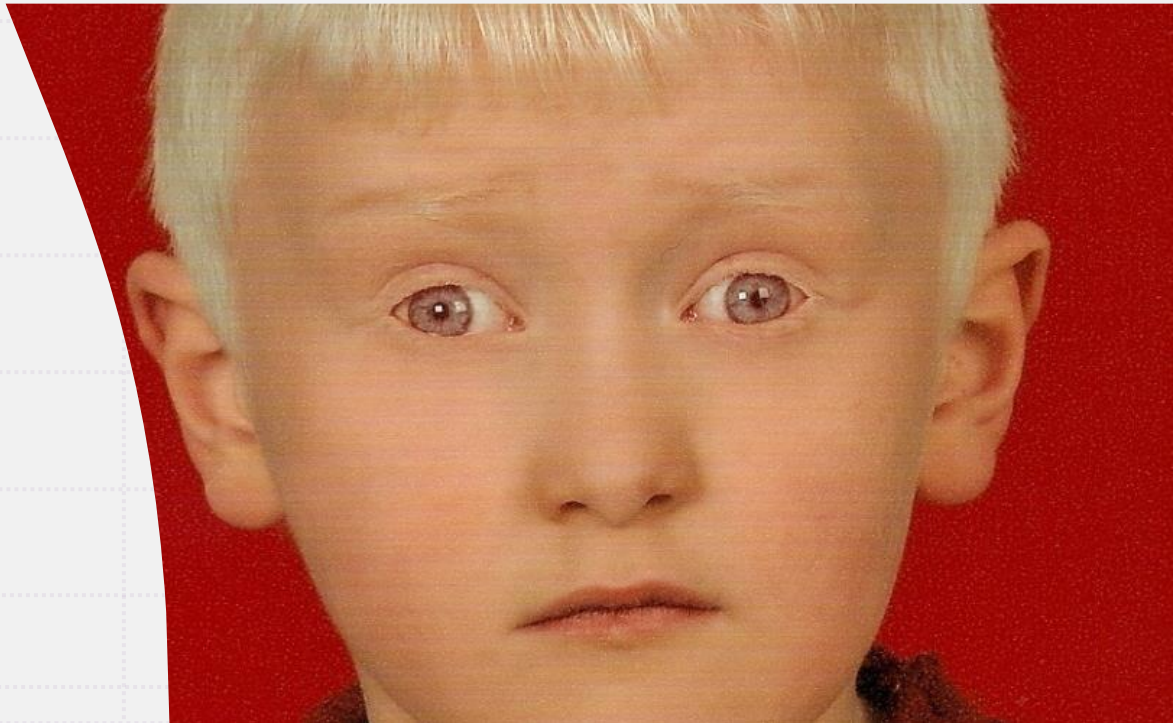




# Clues in hair :Abnormal hair

- Eg:Menkes disease: global delay + hair coloreless , friable,kinky

Low cupper + cerulolasmin



- Gricelli syndrome : Silvery hair

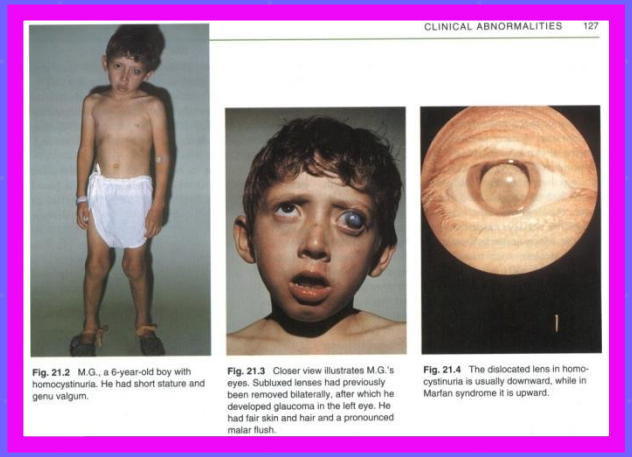
# Clues in the eyes

- **Cataracts :**

-Galactosemia
-Zellweger syndrome
- Lowe syndrome
-Other conditions

- **Dislocated lenses:**

-Homocystinuria,
- Molybdenum co-factor deficiency
-Sulfite oxidase deficiency



- **Retinal degenerative changes**

-peroxisomal disorders
-others

**Cherry red spot :** (mainly lipid storage disease )
-Neimann pick
-Tay sack
-GM1 gangliosidosis
-Sandhoff disease
-Metachromatic leukodystrophy
-mucolipidosis

# Then to summerize

**History and examination** : very important guides towards investigations and diagnosis

If there is no clinical features to suggest a specific diagnosis→ less likely to find a diagnosis

Lab investigations : necessary to reach for a final diagnosis

# Lab investigations

If family history of specific disorder – screen for that disorder

If clue in examination : screen for that disorder

If no hint , what to do ?

# Hearing+ vision assessment

- Should be done **for all** developmentally delayed children

| |
|---|
| Metabolic work up including test for thyroid |
| Karyotyoe :Routine screen ( yield is 3.7% ): it is indicated in the evaluation even in the absence of dysmorphic features |
| Testing for fragile X ( yield 2.6% ):particularly if positive family history of developmental delay |
| Females : frequently affected , may also be considered for testing |

# Advancement in genetic testing

High resolution chromosomal microarray have a diagnostic yield of 15%-20%.

Targeted gene panels has 11-32% diagnostic rate

Whole exome sequencing :has a diagnostic yield of around 40%

whole genome sequencing : 42 %

# EEG

- Not recommended in the routine evaluation if child does not seize

# Neuroimaging

- MRI brain : abn detected in 48-65% of cases

# Identified aetiology

- Traditional tests : diagnostic yield 40% - 60%

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Brain & Development 33 (2011) 810–815

DEVELOPMENT
Official Journal of the Japanese Society of Child Neurology

www.elsevier.com/locate/braindev

Original article

# Profile of developmental delay in children under five years of age in a highly consanguineous community: A hospital-based study – Jordan

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Diagnostic rate **44.5%**

**Abstract**

*Aim:* To assess etiologies and risk factors for global developmental delay (GDD) in children.

*Patients and methods:* Between January 2006 and 2007, a retrospective study was carried out at the Child Neurology Clinic of Jordan University Hospital on all 229 children under five years of age presenting with GDD. To assess risk factors for GDD, 229 age-matched healthy children were included as controls.

# Whole exome sequencing (WES

Powerful tool for etiological discovery in neurodevelopmental disorders

A high-throughput genetic sequencing method that focuses on the protein-coding regions of the genome

While protein-coding genes constitute only 1% of the human genome, they are home to **85 %** of mutations underlying monogenic disorders

Diagnostic rate :**40-60%**

Should be done if all of the previous investigations did no t reveal any cause

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# Global developmental delay and intellectual disability in the era of genomics: Diagnosis and challenges in resource limited areas☆

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Diagnostic rate **44.8%**

ABSTRACT

*Aims:* To report the diagnostic yield of clinical singleton whole exome sequencing (WES) performed among a group of Jordanian children presenting with global developmental delay /intellectual disability (GDD/ID), discuss the underlying identified genetic disorders and the challenges encountered.

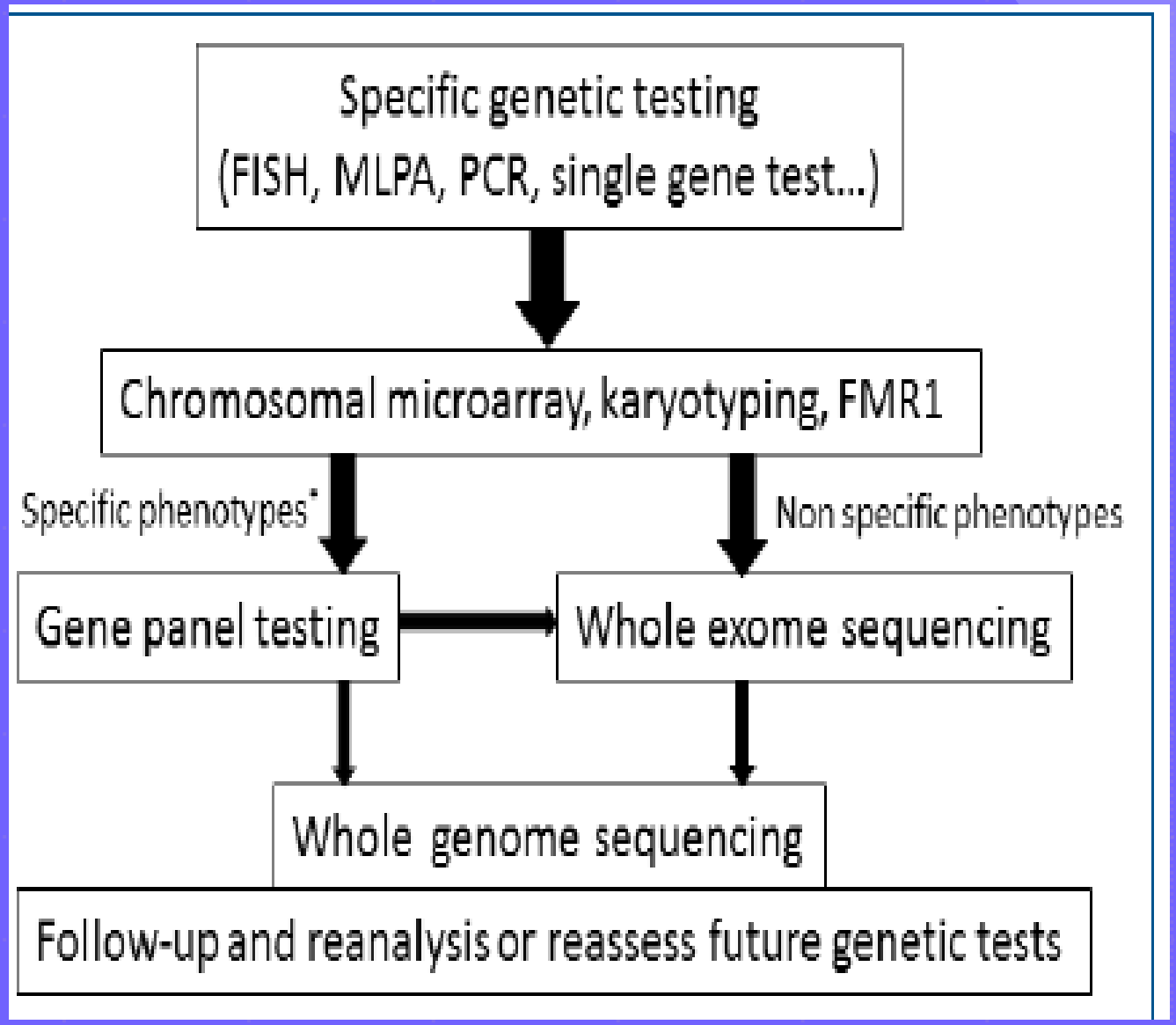
*Patients and methods:* This retrospective medical record review study included 154 children who were diagnosed with GDD/ID at our clinic at Jordan University Hospital between 2016 and 2021, and whose diagnostic work up included WES.

*Results:* Consanguinity among parents was reported in 94/154 (61.0%) patients and history of other affected siblings in 35/154 (22.7%) patients.

Pathogenic and likely pathogenic variants (solved cases) were reported in 69/154 (44.8%) patients, a variant of uncertain significance was reported in 54/154 (35.0%) and a negative result was reported in 31/154 (20.1%) cases.

In the solved cases, autosomal recessive diseases were the most common (33/69; 47.8%). Metabolic disorders were identified in 20/69 (28.9%) patients, followed by developmental and epileptic encephalopathies (9/69; 13.0%) and MECP2 related disorders (7/69; 10.1%). Other single gene disorders were identified in 33/69; 47.8%) patients.

*Conclusion:* This study had several limitations, as it was hospital-based and only including patients who were able to afford the test. Nevertheless, it yielded several important findings. In resource-limited countries, WES may be a reasonable approach. We discussed the challenges that clinicians meet in the context of shortage of resources.


Specific genetic testing
(FISH, MLPA, PCR, single gene test...)
Chromosomal microarray, karyotyping, FMR1
Specific phenotypes*
Non specific phenotypes
Gene panel testing
Whole exome sequencing
Whole genome sequencing
Follow-up and reanalysis or reassess future genetic tests

# Thank you